



EZH2 gene

enhancer of zeste 2 polycomb repressive complex 2 subunit

Normal Function

The *EZH2* gene provides instructions for making a type of enzyme called a histone methyltransferase. Histone methyltransferases modify proteins called histones, which are structural proteins that attach (bind) to DNA and give chromosomes their shape. By adding a molecule called a methyl group to histones (methylation), histone methyltransferases can turn off (suppress) the activity of certain genes, an essential process in normal development. Specifically, the EZH2 enzyme forms part of a protein group called the polycomb repressive complex-2. By turning off particular genes, this complex is involved in the process that determines the type of cell an immature cell will ultimately become (cell fate determination).

Health Conditions Related to Genetic Changes

prostate cancer

Weaver syndrome

More than 30 *EZH2* gene mutations have been identified in people with Weaver syndrome, which involves tall stature, a variable degree of intellectual disability (usually mild), and characteristic facial features. These features can include a broad forehead; widely spaced eyes (hypertelorism); large, low-set ears; a dimpled chin; and a small lower jaw (micrognathia). Some affected individuals have a large head size (macrocephaly). Most of the *EZH2* gene mutations associated with Weaver syndrome change single protein building blocks (amino acids) in the EZH2 enzyme; others insert or delete small amounts of genetic material from the *EZH2* gene, leading to production of an altered EZH2 enzyme. It is unclear how these *EZH2* gene mutations result in the abnormalities characteristic of Weaver syndrome.

cancers

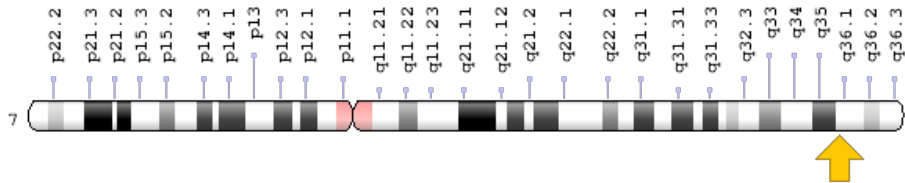
Changes in the *EZH2* gene have been associated with various types of cancers. Mutations of this gene have been identified in cancers of blood-forming tissues (lymphomas and leukemias). These mutations are described as "gain-of-function" because they appear to enhance the activity of the EZH2 enzyme or give the enzyme a new, atypical function. In addition, excessive activity (overexpression) of the *EZH2* gene has been identified in cancerous tumors of the prostate, breast, and other parts of the body. Changes involving the *EZH2* gene likely impair normal control of cell

division (proliferation), allowing cells to grow and divide too fast or in an uncontrolled way and leading to the development of cancer.

Chromosomal Location

Cytogenetic Location: 7q36.1, which is the long (q) arm of chromosome 7 at position 36.1

Molecular Location: base pairs 148,807,372 to 148,884,662 on chromosome 7 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- enhancer of zeste homolog 2 (Drosophila)
- ENX-1
- EZH2_HUMAN
- histone-lysine N-methyltransferase EZH2
- KMT6
- KMT6A
- lysine N-methyltransferase 6

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Chromatin Mechanisms Regulating Gene Expression In Health And Disease
<https://www.ncbi.nlm.nih.gov/books/NBK45032/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28EZH2%5BTI%5D%29+OR+%28enhancer+of+zeste+homolog+2%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

OMIM

- ENHANCER OF ZESTE, DROSOPHILA, HOMOLOG 2
<http://omim.org/entry/601573>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/EZH2ID40517ch7q36.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=EZH2%5Bgene%5D>
- HGNC Gene Family: Lysine methyltransferases
<http://www.genenames.org/cgi-bin/genefamilies/set/487>
- HGNC Gene Family: Myb/SANT domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/532>
- HGNC Gene Family: Polycomb repressive complex 2
<http://www.genenames.org/cgi-bin/genefamilies/set/1347>
- HGNC Gene Family: SET domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/1399>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3527
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/2146>
- UniProt
<http://www.uniprot.org/uniprot/Q15910>

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